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XV. **A PATIENT WITH ADENOSINE KINASE DEFICIENCY DUE TO A NOVEL MUTATION PRESENTING WITH NOVEL DYSMORPHIC AND CARDIAC FINDINGS**
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XVI. **Pompe Hastalarında Enzim Replasman Tedavisine Bağlı Anafilaksi ve Yönetimi:Tek Merkez Deneyim**
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XVIII. **Anatomy of MPS Patients**
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XIX. **Diagnostic workshop: 'Solve the mystery case'**
EZGÜ F. S.
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XX. **Laboratory diagnosis of Mucopolysaccharidosis disorders**
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XXI. **Analysis of hereditary nephrotic diseases through next generation DNA sequencing**
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XXII. **Case Study: Switch from agalsidase alfa to beta**
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XXIII. **Fabry Hastalığı'nda Doğal Seyir**
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XXIV. **Fabry mi? Değil mi?**
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XXV. **Fabry Hastalığı'nda Kalıtım**
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XXVI. **Fabry Hastalığı'nda Tedavi Seçenekleri, Endikasyonlar ve Sorunlar**
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XXVII. **Akut Porfiria Benzeri Periferik Nöropati Gelişen Tirozinemi Tip 1 Olgusu**
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XXVIII. **Genetik Hastalıklarda Yeni Tedaviler**
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XXIX. **Screening of twelve lysosomal storage diseases with LC-MS/MS in Gazi university hospital in Turkey: The firstresults of validation**
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XXX. **Benefficial effects of Modified Atkins Diet in Glygocen Storage Disorder Type IIIa**
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XXXIII. **Next generation DNA sequencing as an initial diagnostic method for congenital defects of glycosylation**
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XXXV. **Natural History in Fabry Disease: Classic vs Late-onset phenotype-Paediatrics**
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XXXVI. **Metabolic and rare diseases: new effective treatments**
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XXXVII. **Population Genetics and Inborn errors of Metabolism**
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XXXVIII. **Hafif Renal Varyant ile Karakterize Pİerson Sendromu: Olgu Sunumu**
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XXXIX. **Fabry Hastalığı**
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XL. **Screening of Twelve Lysosomal Storage Diseases with LC-MS/MS in Gazi University Hospital: The First Results of Validation.**
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XLI. **Cornelia de Lange Syndrome and Glycogen Storage Disease Together in a Patient**
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XLIV. **Familial Hyperphosphatemic Tumoral Calcinosis in an Unusual Site**
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XLV. **Hyperinsulinemic Hypoglycemia: Think of GLUD1 Gene Mutation Leading To Hyperinsulinism/Hyperammonemia (HI/HA) Syndrome**
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XLVII. **Adenylosuccinate Lyase Deficiency in A Turkish Siblings**
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XLVIII. **Novel Mutation in FBP1 Gene Presenting with Recurrent Episodes of Vomiting in A Child**
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XLIX. **Novel Mutation in Two Siblings with Normouricemic Lesch Nyhan Syndrome**
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L. **A Very Rare Disease: Hyperornithinemia-Hyperammonemia-Homocitrullinuria (Hhh) Syndrome**
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LI. **Hyperammonemia Secondary to Mitochondrial HMG-Coa Synthase Deficiency**
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LII. **A Novel Rars2 Mutation in Two Siblings with Microcephaly, Seizures and Liver Involvement**
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LIII. **Pyruvate Carboxylase Ceficiency in A Child with an Early Diagnosis of KetolysisDefect**
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LIV. **Hyperinsulinemic Hypoglycemia: Think of GLUD1 dgene mutation leading to Hyperinsulinemic hyperammonemia (HI/HA syndrome)**
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LV. **Growth Hormone Treatment: Reverses Catabolic Process in Inborn Errors of Metabolism**
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LVI. **Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal storage Diseases**
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LVII. **Screening Approaches and Laboratory Diagnosis in Gaucher Disease**
EZGÜ F. S.
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LVIII. **Case Presentations and Discussion**
EZGÜ F. S.
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LIX. **Insights into Lysosomal Storage Diseases**
EZGÜ F. S.
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LX. **Future of Rare Diseases Research - Advances in Lysosomal Storage Disorders Management**
EZGÜ F. S.
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LXI. **The Practical Aspects of Diagnosis Protocols Implementation**
EZGÜ F. S.
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LXII. **Safety and tolerability of SOBI003 in pediatric MPS IIIA patients key study design features of the ongoing first-in-human study**
Broijersen A., Dalen P., Ezgu F. S., Huledal G., Lindqvist D., Wiken M., Harmatz P.
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LXIII. **ICV-administered tralesinidase alfa (BMN 250 NAGLU-IGF2) is well-tolerated and reduces heparan sulfate accumulation in the CNS of subjects with Sanfilippo syndrome type B (MPS IIIB)**
Cleary M., Muschol N., Luz Couce M., Harmatz P., Lee J., Lin S., OKUR İ., Ezgu F. S., Peters H., Villarreal M. S., et al.
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LXIV. **Natural history data for young subjects with Sanfilippo syndrome type B (MPS IIIB)**
Villarreal M. S., OKUR İ., Cleary M., Lopez M. J. d. C., Harmatz P., Lee J., Lin S., Couce M. L., Muschol N., Peters H., et al.

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LXV. **Once every 4 weeks-2 mg/kg of pegunigalsidase alfa for treating Fabry disease Preliminary results of a phase 3 study**
Holida M. D., Bernat J., Longo N., Goker-Alpan O., Wallace E., Schiffmann R., Deegan P., Khan N., Tondel C., Eyskens F., et al.
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LXVI. **Pompe hastalığının tedavisinde güncel gelişmeler**
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Korkut Yaltkaya XIII. Klinik Nörofizyoloji Sempozyumu, Antalya, Türkiye, 21 - 23 Aralık 2018

LXVII. **Fabry hastalığının tedavisi ve seçenekler**
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Korkut Yaltkaya XIII. Klinik Nörofizyoloji Sempozyumu, Antalya, Türkiye, 21 - 23 Aralık 2018

LXVIII. **Gaucher Disease, is it still a diagnostic challenge after 30 years of clinical experience. Best Practices**
EZGÜ F. S.
Lysosomal Storage Disease Expert Meeting, Beirut, 2018, 15 Aralık 2018

LXIX. **Best Practices from Turkey Focus on Awareness, Access Patient Journey**
EZGÜ F. S.
"Second Step Towards Enhancing LSD Patient's Journey"The second Rare Diseases Out Loud Advisory Board MeetingLysosomal Storage Disorders, Beirut, 2018, 14 Aralık 2018

LXX. **Gaucher Disease: from Diagnosis to Treatment**
EZGÜ F. S.
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LXXI. **RDs International vs local reimbursement guidelines**
EZGÜ F. S.
"Second Step Towards Enhancing LSD Patient's Journey"The second Rare Diseases Out Loud Advisory Board MeetingLysosomal Storage Disorders, Beirut, 2018, 14 Aralık 2018

LXXII. **Forum conclusion and Take Home Messages**
EZGÜ F. S.
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LXXIII. **LSDs Landscape**
EZGÜ F. S.
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LXXIV. **Turkish experience (Sharing Best Practices): ''Moving from Diagnosis, Awareness to Access'**
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LXXV. **LSDs landscape From Diagnosis to treatment with special focus on Gaucher Disease**
EZGÜ F. S.
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LXXVI. **RAR2mutation in two siblingswith microcephaly,seizures and liver involvement**
EMİNOĞLU F. T., s s., gök t., EZGÜ F. S., İNCİ A., TÜMER L.
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LXXVII. **Respiratory system involvement of 41 Mucopolysaccaridosis patients with the evaluation of KL-6, SPA and SPD levels**
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LXXVIII. **Closing Day 1**
EZGÜ F. S.

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EZGÜ F. S.
4th Gaucher Disease Regional Forum, 2 - 03 Kasım 2018

LXXX. **Wrap Up**
EZGÜ F. S.
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LXXXI. **Advances in Gaucher Disease Management: Available Choices and Direction of Future Research**
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LXXXII. **Gaucher Disease Is it still a diagnostic challenge after 30 years of clinical experience?**
EZGÜ F. S.
4th Gaucher Disease Regional Forum, İstanbul, 2 - 03 Kasım 2018

LXXXIII. **UNIQUE CLINICAL AND MOLECULAR FINDINGS IN LARGE COHORT OF PATIENTS WITH GAUCHER DISEASE FROM TURKEY**
Akay Tayfun G., OKUR İ., BİBEROĞLU G., TÜMER L., İNCİ A., Küçükcongar A., Hasanoğlu A., EZGÜ F. S.
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LXXXIV. **Diagnosis and Testing**
EZGÜ F. S.
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LXXXV. **FMF'e genetik yaklaşım**
EZGÜ F. S.
VI. Çocuk genetik hastalıkları sempozyumu, Gaziantep, 2018, Türkiye, 19 Ekim 2018

LXXXVI. **Association of hereditary focal and segmental glomerulosclerosis with Bartter syndrome: a case report**
YAZICIOĞLU B., BÜYÜKKARAGÖZ B., ALPMAN B. N., IŞIK GÖNÜL İ., EZGÜ F. S., BUYAN N., BAKKALOĞLU EZGÜ S. A.
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LXXXVII. **Complement factor B mutation in atypical hemolytic uremic syndrome: a case presentation**
ÇELEBİ TAYFUR A., ÇALTIK YILMAZ A., İNAN Y., KARAOKUR E., BÜYÜKKARAGÖZ B., KOÇAK M., EZGÜ F. S.
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LXXXVIII. **Multidisipliner bir hastalık: Deneyimler ışığında Fabry**
EZGÜ F. S.
35. Ulusal nefroloji, hipertansiyon, diyaliz ve transplantasyon kongresi, 2018, Antalya, Türkiye, 3 - 07 Ekim 2018

LXXXIX. **ICV-administered BMN 250 (NAGLU-IGF2) is Well Tolerated and Reduces Heparan Sulfate Accumulation in the CNS of Subjects with Sanfilippo Syndrome Type B (MPS IIIB)**
Muschol N., Cleary M., Couce M., Harmatz P., Lee J., Lin S. P., Okur İ., Ezgu F. S., Peters H., Villarreal M., et al.
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XC. **Natural History Data for Young Subjects with Sanfilippo Syndrome Type B (MPS IIIB)**
Okur I., Cleary M., Lopez d. C. M., Harmatz P., Lee J., Lin S. P., Couce M., Muschol N., Peters H., Villarreal M., et al.
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XCI. **Çocuklarda spontan pnömotoraks nedeni olarak marfan sendromu**
RAMASLI GÜRSOY T., ŞİŞMANLAR EYÜBOĞLU T., ASLAN A. T., EZGÜ F. S.
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XCII. **Glycogen storage disease type 9: Insidious onset,mild form**
TÜMER L., İNCİ A., OKUR İ., BİBEROĞLU G., EZGÜ F. S.
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XCIII. **Determination of succinylacetone in dried blood spot: preliminary results of our laboratory**
BİBEROĞLU G., TÜMER L., OKUR İ., EZGÜ F. S., İNCİ A.
SSIEM, 4 - 07 Eylül 2018

XCIV. **An early diagnosis cerebretendinous xanthomatosis in a patient at the age of 15 years**
İNCİ A., BİBEROĞLU G., OKUR İ., TÜMER L., EZGÜ F. S.
SSIEM, 4 - 07 Eylül 2018

XCV. **Respiratory system involvement of mucopolysaccaridosis patients with the evaluation of KL-6, SPA and SPD levels**
İNCİ A., OKUR İ., YILMAZ-DEMİRTAŞ C., BİBEROĞLU G., aslan A. T., EZGÜ F. S., TÜMER L.
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XCVI. **The clinical evaluation of Fabry patientswith Mainz severity score index and DS3 score**
OKUR İ., İNCİ A., bütün s., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
SSIEM, 4 - 07 Eylül 2018

XCVII. **Laboratory Diagnosis of MPS Disorders**
EZGÜ F. S.
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XCVIII. **Next generation sequencing in the diagnosis of metabolic diseases**
EZGÜ F. S.
Organelle disease revisited, 2018, Antwerp, 18 Mayıs 2018

XCIX. **Erken Başlangıçlı Lizozomal Asit Lipaz Eksikliğinde Enzim Replasman Tedavisi Sonuçları**
EZGÜ F. S.
VI. Uluslararası katılımlı Lizozomal Hastalıklar Kongresi, Türkiye, 11 - 15 Nisan 2018

C. **Natural history data for young subjects with Sanfilippo Syndrome Type B (MPS IIIB)**
OKUR İ., Cleary M., de Castro Lopez M. J., Harmatz P., Lees J., Lin S., Luz Couce M., Muschol N., Peters H., Solano Villarrea M., et al.
40th Annual Meeting of the Society-for-Inherited-Metabolic-Disorders (SIMD), California, Amerika Birleşik Devletleri, 11 - 14 Mart 2018, cilt.123, ss.255-256

CI. **RENAL INVOLMENT IN FABRY DİSEASE**
İNCİ A., BİBEROĞLU G., PAŞAOĞLU Ö. T., TÜMER L., PAŞAOĞLU H., EZGÜ F. S.
14 th middle east metabolic group (MEMG) meeting Athens GREECE, Atina, Yunanistan, 9 - 11 Şubat 2018

CII. **Natural history data for young subjects with Sanfilippo syndrome type B (MPS IIIB)**
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CIII. **Düşündüğüm metabolik hastalığa nasıl tanı koyabilirim?**
EZGÜ F. S.
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CIV. **Atipik hemolitik üremik sendromda kompleman faktör H mutasyonu: Vaka sunumu**
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